Kidney: Renal cell carcinoma with t(X;1)(p11;p34) SFPQ/TFE3

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Abstract

Review on Renal cell carcinoma with t(X;1)(p11;p34) SFPQ/TFE3, with data on clinics, and the genes involved.

Keywords
Renal cell carcinoma; chromosome X; chromosome 1; SFPQ; TFE3; MiT family

Identity
Must not be confused with the t(X;1)(p11.2;q21), also found in renal cell carcinoma.

Classification
Xp11 translocation renal cell carcinoma (RCCs) harbor gene fusions involving TFE3 transcription factor. The t(6;11) RCCs harbor a specific MALAT1 (Alpha) - TFEB gene fusion. TFEB and TFE3 belong to the same MiT subfamily of transcription factors. Because of similarities at the clinical, morphologic, immunohistochemical, and genetic levels, the Xp11 translocation RCCs and t(6;11) RCCs are currently grouped together under the category of MiT family translocation renal cell carcinoma.

Clinics and pathology

Disease

t(X;1)(p11.2;p34) is found in a subset of Xp11 translocation RCC, median age 36 years. These tumors often have papillary architecture and clear cells with subnuclear vacuoles mimicking clear cell papillary RCC, or a nested clear cell appearance mimicking clear cell RCC.

Phenotype / cell stem origin

The identical gene fusion may be found in Xp11 translocation PEComas and Melanotic Xp11 translocation cancers, which differ from the RCC in that they are PAX8 negative and Cathepsin K positive by immunohistochemistry.

Genes involved and proteins

TFE3 (transcription factor E3)

Location

Xp11.23
Protein
Contains a transcriptional activation domain, a helix-loop-helix, and a leucine zipper; member of the basic helix-loop-helix family (b-HLH) of transcription factors.

**SFPQ (PTB-associated splicing factor)**

**Location**
1p34.3

**Protein**
Contains RNA binding domains; involved in pre-mRNA splicing; form complexes with DNA topoisomerase I.

Result of the chromosomal anomaly

**Hybrid Gene**

**Description**
5’ SFPQ - 3’ TFE3

**Fusion Protein**

**Description**
N-term SFPQ and most of it fused to the DNA binding domains of TFE3 (excluding the acidic transcriptional activation domain, including the C-term helix-loop-helix, and the leucine zipper); no TFE3-SFPQ reciprocal transcript, as the der(X)

(t(X:1) is missing; the normal TFE3 transcript is found.

References


This article should be referenced as such: